Bionano Solve v3.8 Release Notes

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bionano

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Revision History

REVISION	NOTES
A	Initial release of document.

Introduction

This document describes the release of Bionano Solve 3.8. This is an overview of the fixes and improvements of the Bionano Solve analysis tools and pipelines to provide a better understanding of the impact of moving to this version of the software. Should any questions arise, please contact support@bionano.com.

Bionano Tools and Bionano Solve are combined and branded as Bionano Solve. Bionano Solve is installed on Saphyr Compute, Bionano Compute, and Bionano Access Servers before server shipment and installation.

Bionano Solve (folder “tools”) is located at the /home/bionano directory on the Compute server. The folder contains a collection of tools and scripts. Each individual tool is versioned independently. These tools together perform bioinformatics analyses on the Compute server.

Compatibility

Bionano Solve 3.8 is compatible with Bionano Access 1.8 only.

References

Visit https://bionano.com/software-and-data-analysis-support-materials// for file format specifications and Theory of Operation documents.

Support for new reference genomes

Telomere-to-Telomere Consortium Reference

- Implemented system level support for the T2T-CHM13v2.0 reference genome
- Integrated T2T-CHM13v2.0 gene annotation
- Created Bionano Control Database versions for *de novo* assembly and Rare Variant Analysis pipeline

mm39 Reference

- Implemented system level support for the mouse mm39/GRCm39 reference genome
- Integrated mm39 gene annotation
- Created Bionano Control Database versions for *de novo* assembly and Rare Variant Analysis pipeline

Singularity

- Adopted Singularity for dependency management of the Solve pipeline (replacing Docker)
- Provides method for portable, consolidated dependency management across supported Compute hardware
- Designed for ease-of-use on high-performance computing (HPC) systems

Provides improved security for customers by removing requirements for running with elevated or root user privileges

Improved support for standard genomics file formats

- Introduced OGM BAM output (binary version of the Sequence Alignment/Map format) for molecule-to-reference alignments.
- Updated OGM Variant Call File (VCF) with improved representations of variants and quality filters.
- Updated OGM VCF to include Solve AOH calls for *de novo* and guided assembly – constitutional analyses.
- Updated OGM VCF to contain clustered variant calls only to remove redundant calls.
  - Translocations not included in clustering for initial release.
- Enables import of OGM data into VIA as well as any standards-compliant genomics software that supports BAM and VCF.

Control database

- Added 394 DLE-1 samples to human control database.
- Complete reanalysis of control databases for *de novo* assembly and Rare Variant Analysis for hg19 and hg38.
- Introduction of control database for T2T-CHM13v2 reference genome.

Gene annotations

- Updated gene annotation used by Variant Annotation Pipeline to harmonize gene annotations used by Access and VIA.
- Updated gene annotation to latest RefSeq builds for hg38 and hg19.
- Introduced gene annotation for T2T-CHM13v2.0 and mm39.
- Streamlined gene annotation to provide a single, high quality, current annotation for human analyses while allowing use of custom annotations if desired.

Y-PAR Masked Human References

- Added versions of human reference genomes that mask out the pseudoautosomal regions (PAR) on chromosome Y similar to approaches used in many Next-Generation Sequencing (NGS) analysis workflows.
- This is done to address the sequence homology in these regions with the corresponding regions on chromosome X which can interfere with map and molecule alignments to the reference. Masking of these regions has been shown to improve structural and copy number variant calling for genes such as *CRLF2* that are in or near the region.
- Masked references are provided as options for hg19 and hg38 and by default for T2T-CHM13v2.0. For best results with SV detection performance and annotation with the Bionano control databases, the masked references are recommended for hg19 and hg38.

Stable Region Analysis

- Incorporated analysis of stable regions as a quality control step.
- Initially implemented in the EnFocus™ FSHD and EnFocus™ Fragile X applications, assessment of stable regions is now included in all human whole genome analysis pipelines (*de novo* assembly and Rare Variant Analysis).
- Output included in informatics report

Known Issues and Limitations

- CNV calls for mouse reference mm10 have confidence scores of zero. This will be corrected in a future release.
- Translocation calls are not clustered in the VCF as are other variant types. This is due to a limitation of the reporting of supporting alignments in the XMAP and will be corrected in a future release.
- VCF representation of deletion calls produced by the RVA pipeline uniformly list genotype as heterozygous for short term compatibility with downstream analysis tools.
- Control databases were generated using the Y-PAR masked versions of hg19 and hg38. These databases can be used to annotate data analyzed using the unmasked versions of the reference genome, but SV calls in Y PAR1 and PAR2 will show as having 0% match in the control db.

Technical Assistance

For technical assistance, contact Bionano Technical Support.

You can retrieve documentation on Bionano products, SDS's, certificates of analysis, frequently asked questions, and other related documents from the Support website or by request through e-mail and telephone.

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